CYP21A2 gene

cytochrome P450 family 21 subfamily A member 2

Normal Function

The *CYP21A2* gene provides instructions for making an enzyme called 21-hydroxylase, which is part of the cytochrome P450 family of enzymes. Cytochrome P450 enzymes are involved in many processes in the body, such as assisting with reactions that break down drugs and helping to produce cholesterol, certain hormones, and fats (lipids).

The 21-hydroxylase enzyme is found in the adrenal glands, which are located on top of the kidneys and produce a variety of hormones that regulate many essential functions in the body. 21-hydroxylase plays a role in producing hormones called cortisol and aldosterone. Cortisol helps maintain blood sugar levels, protects the body from stress, and suppresses inflammation. Aldosterone is sometimes called the salt-retaining hormone because it regulates the amount of salt retained by the kidneys. The retention of salt affects fluid levels in the body and blood pressure.

Health Conditions Related to Genetic Changes

21-hydroxylase deficiency

More than 100 mutations in the *CYP21A2* gene have been found to cause 21-hydroxylase deficiency. Some of these mutations result from an exchange of genetic material between the *CYP21A2* gene and a similar but nonfunctional piece of DNA called a pseudogene, which is located very close to the *CYP21A2* gene on chromosome 6. This type of DNA exchange is called a gene conversion. The genetic material from the pseudogene contains errors that, when introduced into the *CYP21A2* gene, disrupt the way the gene's instructions are used to make a protein. Other mutations that cause 21-hydroxylase deficiency change single protein building blocks (amino acids) in the 21-hydroxylase enzyme or delete or insert pieces of DNA in the *CYP21A2* gene.

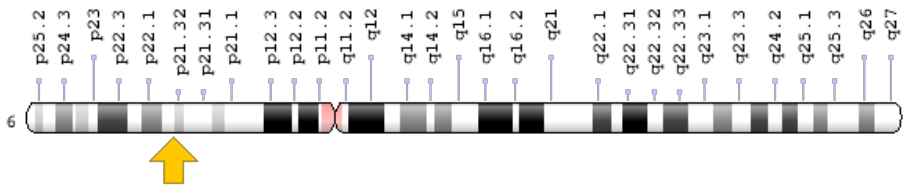
Researchers have described three forms of 21-hydroxylase deficiency. Individuals with a form of the disorder called the salt-wasting type have *CYP21A2* mutations that result in a completely nonfunctional enzyme. People with the simple virilizing type of this condition have *CYP21A2* gene mutations that allow the production of low levels of functional enzyme. Individuals with the non-classic type of this disorder have *CYP21A2* mutations that result in the production of reduced amounts of the enzyme, but more enzyme than any of the other types. All types of 21-hydroxylase deficiency interfere with the production of cortisol and aldosterone. The substances that are usually used to form these hormones instead build up in the adrenal glands and

are converted to androgens, which are male sex hormones. The excess production of androgens leads to abnormalities of sexual development in people with 21-hydroxylase deficiency.

Chromosomal Location

Cytogenetic Location: 6p21.33, which is the short (p) arm of chromosome 6 at position 21.33

Molecular Location: base pairs 32,038,316 to 32,041,670 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CA21H
- CAH1
- CP21A_HUMAN
- CPS1
- CYP21
- CYP21B
- Cytochrome P450 Family 21 Subfamily A Polypeptide 2
- Cytochrome P450 XXI
- cytochrome P450, family 21, subfamily A, polypeptide 2
- cytochrome P450, subfamily XXIA (steroid 21-hydroxylase, congenital adrenal hyperplasia), polypeptide 2
- Cytosteroid 21-Monooxygenase
- P450c21B
- steroid 21-hydroxylase
- steroid 21-monooxygenase

Additional Information & Resources

Educational Resources

- Endocrinology: An Integrated Approach (first edition, 2001): Congenital adrenal hyperplasia (CAH) - CYP21A2 deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK26/box/A593/

GeneReviews

- 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia
  https://www.ncbi.nlm.nih.gov/books/NBK1171

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28CYP21A2%5BTIAB%5D%29+OR+%28%28CAH1%5BTIAB%5D%29+OR+%28CYP21%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY
  http://omim.org/entry/201910

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CYP21A2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CYP21A2%5Bgene%5D
- HGNC Gene Family: Cytochrome P450 family 21
  http://www.genenames.org/cgi-bin/genefamilies/set/1011
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2600
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1589
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  http://www.uniprot.org/uniprot/P08686

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